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ADVANCING SURGICAL CARE

CASE REPORTS

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Staged sternal distraction for early thoracic expansion in Shwachman–Diamond syndrome

Shwachman–Diamond syndrome (SDS) is a rare inherited multisystem disorder with variable skeletal involvement; severe neonatal thoracic dystrophy is uncommon but can cause thoracic insufficiency and mimic Jeune syndrome. We report staged sternal distraction in a 5-month-old infant with a severely constricted thorax, pulmonary hypoplasia, refractory pulmonary hypertension, tracheostomy-dependent respiratory failure and an iatrogenic posterior tracheal wall injury. Subsequent trio long-read genome sequencing identified compound heterozygous pathogenic variants in *SBDS*, c.183_184delinsCT (p.Lys62Ter), inherited paternally, and c.258+2T>C, inherited maternally, establishing a molecular diagnosis of SDS. At presentation, the chest circumference measured 36cm, compared with an age-expected value of 41cm. The patient underwent staged median sternotomy distraction followed by stabilisation with custom fibular allograft struts. At approximately 3 months, chest circumference had increased to 41cm, peak inspiratory pressures had decreased from 40–50cm H_2O to the mid-20cm H_2O range, pulmonary hypertension was no longer evident on echocardiography, and the tracheal injury had healed. At 6 months, he remained on home mechanical ventilation, with peak inspiratory pressures in the upper 20cm H_2O range. To our knowledge, operative thoracic expansion specifically for SDS-associated thoracic insufficiency has not previously been described. In carefully selected infants with severe thoracic insufficiency who are not yet candidates for conventional thoracic reconstruction, staged sternal distraction may serve as an interim strategy to achieve meaningful thoracic expansion.

Keywords: Shwachman–Diamond syndrome; Thoracic insufficiency syndrome; Thoracic dystrophy; Sternal distraction; Early thoracic expansion

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Background

Shwachman–Diamond syndrome (SDS) is a rare inherited multisystem disorder characterised by bone marrow dysfunction, exocrine pancreatic dysfunction, growth impairment and variable skeletal abnormalities. The most common molecular form is associated with biallelic pathogenic variants in *SBDS*.[1] The skeletal phenotype is heterogeneous and can include metaphyseal dysplasia, delayed ossification, rib abnormalities and thoracic narrowing.[2] Rare infants present with severe thoracic dystrophy and respiratory failure caused by markedly short ribs and a constricted chest.[3–7] These presentations can closely resemble asphyxiating thoracic dystrophy (Jeune syndrome); in one genetically confirmed SDS cohort, 4 of 11 patients had initially been diagnosed with Jeune syndrome because of severe thoracic dystrophy.[6]

Thoracic insufficiency syndrome describes the inability of the thorax to support normal respiration or lung growth.[8,9] Most operative experience for congenital thoracic restriction has been reported in Jeune syndrome and related phenotypes. Techniques include vertical expandable prosthetic titanium rib (VEPTR) implantation, lateral thoracic expansion, sternoplasty, external thoracic expansion, progressive internal sternal distraction and structural bone-graft expansion thoracoplasty.[10–17] Very young infants with severe physiological compromise may not yet be candidates for conventional reconstruction because of small body size, skeletal immaturity or concern that abrupt expansion will not be tolerated.

We report staged median sternotomy distraction in a 5-month-old infant with severe thoracic insufficiency whose molecular diagnosis was established postoperatively as SDS. Thoracic expansion was achieved through staged sternal distraction followed by stabilisation with custom fibular allograft struts. The case illustrates both an uncommon thoracic presentation of SDS and a potential rescue strategy for infants with progressive cardiopulmonary compromise before conventional thoracic reconstruction is feasible.

Case history

A male infant was delivered at 39 3/7 weeks of gestation and remained hospitalised in the

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neonatal intensive care unit for approximately 5 months because of severe thoracic insufficiency and chronic respiratory failure requiring tracheostomy and mechanical ventilation. Peak inspiratory pressures were 40–50cm H_2O, reflecting severe restrictive thoracic physiology. At transfer, he had a markedly constricted thoracic cage with pulmonary hypoplasia, pulmonary hypertension refractory to sildenafil, gastrostomy dependence and persistent methicillin-resistant *Staphylococcus aureus* (MRSA) colonisation. The thoracic phenotype had initially been considered in the differential diagnosis of short-rib thoracic dysplasia, but prior genetic evaluation had not established a molecular diagnosis. Subsequent trio long-read genome sequencing identified two pathogenic *SBDS* variants in trans: c.183_184delinsCT (p.Lys62Ter), inherited paternally, and c.258 +2T>C, inherited maternally, establishing a molecular diagnosis of Shwachman–Diamond syndrome. Chest circumference at the nipple line measured 36cm (Figure 1a), compared with an age-expected circumference of approximately 41cm.[18]

Preoperative evaluation included chest computed tomography, transthoracic echocardiography and comprehensive airway assessment. Microlaryngoscopy and bronchoscopy demonstrated a large ulcerative injury of the posterior tracheal wall 2–3cm proximal to the carina, attributed to chronic tracheostomy cuff compression, with associated tracheomalacia and tracheal dilation but no full-thickness disruption. Because of the injury, airway manipulations required endotracheal intubation distal to the lesion under direct bronchoscopic visualisation.

The operative plan was developed collaboratively by paediatric surgery, orthopaedic surgery, cardiothoracic surgery, otolaryngology and paediatric intensive care teams. The primary objective was to increase thoracic volume and support ongoing lung growth. Given the patient's medical complexity and concern regarding tolerance of abrupt expansion, a staged median sternotomy distraction approach was selected to achieve progressive thoracic enlargement while preserving future reconstructive options. An initial 2-cm sternal distraction, followed approximately 2 weeks later by expansion to 4cm with biologic stabilisation, was planned. A single-stage distraction from 0 to 4cm was not feasible because the overlying soft tissues would not permit tension-free closure, and staging allowed assessment of physiologic tolerance before further expansion.

Stage 1 (sternotomy and initial distraction)

After bronchoscopic confirmation that the endotracheal tube was positioned distal to the tracheal injury, the tracheostomy stoma was packed with xeroform gauze, perioperative vancomycin was administered, and a skin bridge was preserved between the tracheostomy and sternotomy incision. A standard median sternotomy was performed, and a HyperDrive™ distractor, 20mm, 36-hole (KLS Martin Group, Jacksonville, FL, USA) was selected. Because the sternum was largely cartilaginous, adequate screw purchase could not be obtained; the distractor was therefore secured to each hemisternum using interrupted 2-0 Ethibond sutures (Ethicon, Inc., Somerville, NJ, USA) passed through the sternum. Two centimetres of distraction were achieved without haemodynamic instability, and the patient returned to the intensive care unit intubated (Figure 2a).

Stage 2 (fixation with custom fibular allograft struts)

Approximately 2 weeks later, the patient returned to the operating room for further thoracic expansion. Following repeat airway assessment, the previous incision was reopened, the distractor was removed and the sternum was distracted to 4cm superiorly and 3cm inferiorly. A 10-cm structural fibular shaft allograft (MTF Biologics, Edison, NJ, USA) was divided into two segments and contoured into low-profile structural struts with U-shaped ends designed to seat securely against the medial hemisterna while minimising anterior projection beneath the skin. Two #2 FiberWire sutures (Arthrex, Inc., Naples, FL, USA) were passed through the medullary canal of each graft and used to secure the struts to the cartilaginous sternum (Figure 2b). A 4-cm strut was positioned superiorly and a 3-cm strut inferiorly, maintaining the distracted sternum in its expanded configuration and providing stable fixation without cortical screw purchase. Bilateral Blake pleural drains were placed, and the overlying soft tissues were mobilised to permit tension-free closure. The wound was closed in layers using 3-0 Vicryl (Ethicon), 4-0 Monocryl (Ethicon) and Dermabond adhesive (Ethicon).

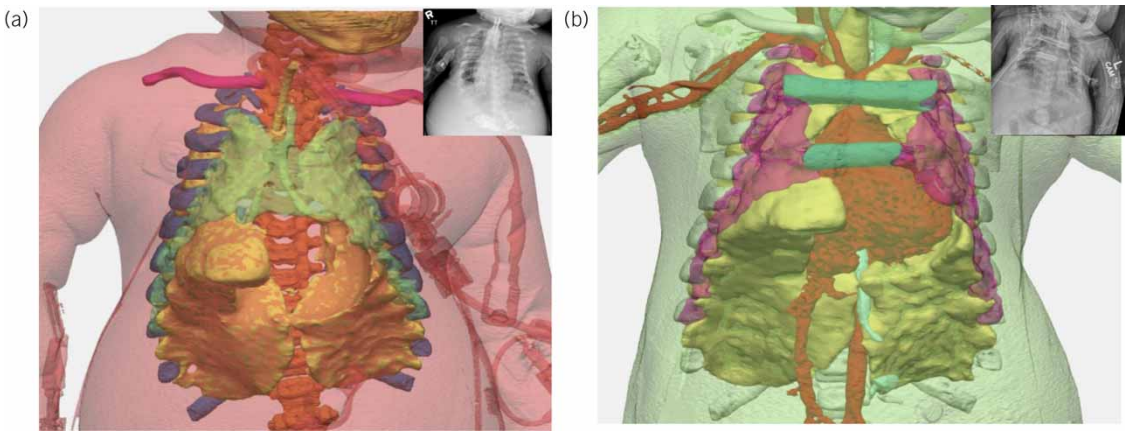


Figure 1 Three-dimensional computed tomography reconstructions of the thorax. (a) Preoperative reconstruction demonstrating a severely constricted thoracic cage with foreshortened, horizontally oriented ribs. The corresponding chest radiograph is shown in the upper right-hand corner. Orange = vertebral column; blue = ossified ribs; green = costal cartilage; yellow = lungs. (b) Postoperative reconstruction demonstrating widened sternal separation maintained by the fibular allograft struts and expansion of the thoracic cage compared with the preoperative study. The corresponding postoperative chest radiograph is shown in the upper right-hand corner.

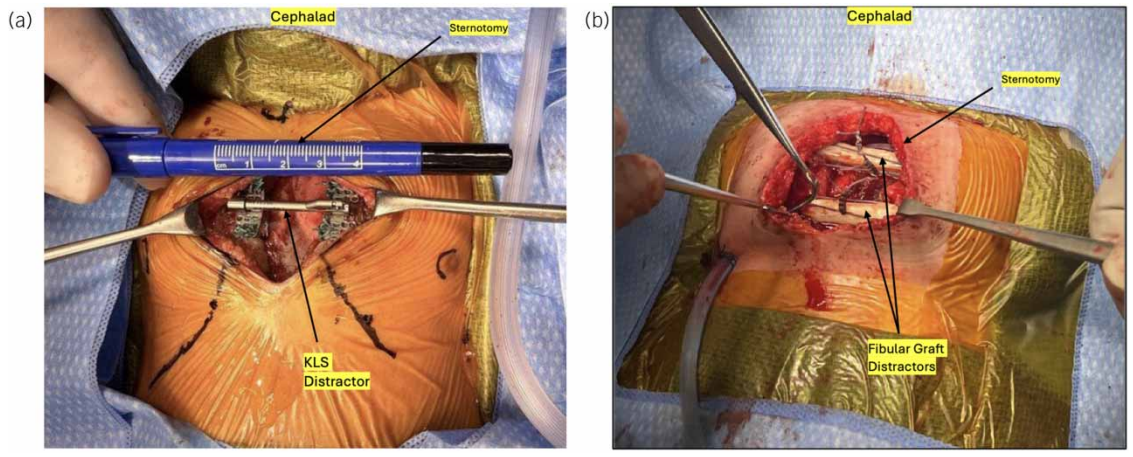


Figure 2 Intraoperative stages of thoracic expansion and definitive stabilisation. (a) Stage 1: HyperDrive™ distractor (KLS Martin Group, Jacksonville, FL, USA) spanning the median sternotomy, with approximately 2cm of initial sternal distraction achieved using suture fixation to the cartilaginous hemisterna. (b) Stage 2: definitive stabilisation with custom-contoured superior and inferior fibular allograft struts spanning the distracted sternum. The U-shaped graft ends seat against the medial hemisterna, and preloaded #2 FiberWire sutures pass through the medullary canal of each graft to secure the struts without cortical screw purchase.

Recovery following Stage 1 was notable for a transient increase in pulmonary artery pressure. Pulmonary hypertension subsequently improved; inhaled nitric oxide was discontinued, and it was no longer evident on follow-up echocardiography. During the postoperative course, tracheal cultures grew MRSA and *Stenotrophomonas* species, and the patient developed *Enterococcus faecalis* bacteraemia; all were treated with appropriate antimicrobial therapy. Following Stage 2, the patient remained intubated for approximately 48h before returning to his tracheostomy. The tracheostomy tube was positioned distal to the area of tracheal injury, just proximal to the carina, with cuff inflation limited to 3ml based on serial bronchoscopic assessments. The posterior tracheal lesion progressively healed over the ensuing weeks.

At approximately 3 months after Stage 2, chest circumference had increased from 36cm preoperatively to 41cm, within the age-expected range (Figure 1b). The fibular allograft construct remained stable and tolerated routine chest physiotherapy and vibration vest treatments without displacement or loss of fixation. There was no evidence of graft infection, exposure or overlying skin breakdown. Although the patient remained dependent on chronic mechanical ventilation, peak inspiratory pressures decreased from 40–50cm H_2O preoperatively to the mid-20cm H_2O range. At 6-month follow-up, he remained on home mechanical ventilation with peak inspiratory pressures in the upper 20cm H_2O range. Formal pulmonary function testing was not feasible because of his age and ventilator dependence; chest dimensions, ventilatory pressure requirements and serial echocardiography therefore served as practical objective surrogates of early physiologic response.

Discussion

The postoperative molecular diagnosis changes the aetiologic classification of this case, but not the mechanical problem that prompted surgery. SDS is a multisystem disorder with a variable skeletal phenotype, not a short-rib thoracic dysplasia/ciliopathy. Nevertheless, severe thoracic dystrophy is a recognised SDS presentation and can be difficult to distinguish phenotypically from Jeune syndrome during infancy.[2–7] In this patient, the clinically relevant consequence was profound thoracic restriction causing pulmonary hypoplasia, chronic ventilator dependence, pulmonary hypertension and progressive airway morbidity.

The principal clinical lesson is that consideration of thoracic expansion should be driven by the severity and trajectory of thoracic insufficiency rather than by a specific diagnostic label alone. Before referral, the patient had spent approximately 5 months in intensive care with high-pressure ventilatory requirements, refractory pulmonary hypertension and an acquired posterior tracheal injury. Although a single case cannot establish an optimal timing threshold, persistent cardiopulmonary compromise in an infant with a mechanically constricted thorax should prompt early multidisciplinary evaluation for expansion rather than automatic deferral solely because of young age or small size.

This technique should be considered a highly selected rescue or bridge strategy rather than a standard alternative to established thoracic expansion procedures. Potential candidates would include infants with life-threatening thoracic insufficiency despite optimised medical support, anatomy in which anterior expansion can meaningfully increase thoracic volume, and skeletal immaturity or small body size that limits conventional hardware or reconstruction. Staging may be useful when full acute expansion could be poorly tolerated physiologically or would prevent tension-free soft-tissue closure. The approach requires coordinated expertise from paediatric surgery, cardiothoracic surgery, orthopaedics, airway, anaesthesia and critical care.

The staged median sternotomy approach allowed progressive thoracic enlargement while accommodating the patient's medical complexity. The largely cartilaginous sternum precluded reliable screw fixation, so definitive stabilisation was achieved with custom-contoured fibular allograft struts secured by transsternal suture fixation (Figure 3). The strategy is related to previously reported sternotomy-based procedures, including sternoplasty and progressive internal sternal distraction,[14–16] and to structural bone-graft expansion thoracoplasty.[17] However, we identified no previous report of operative thoracic expansion

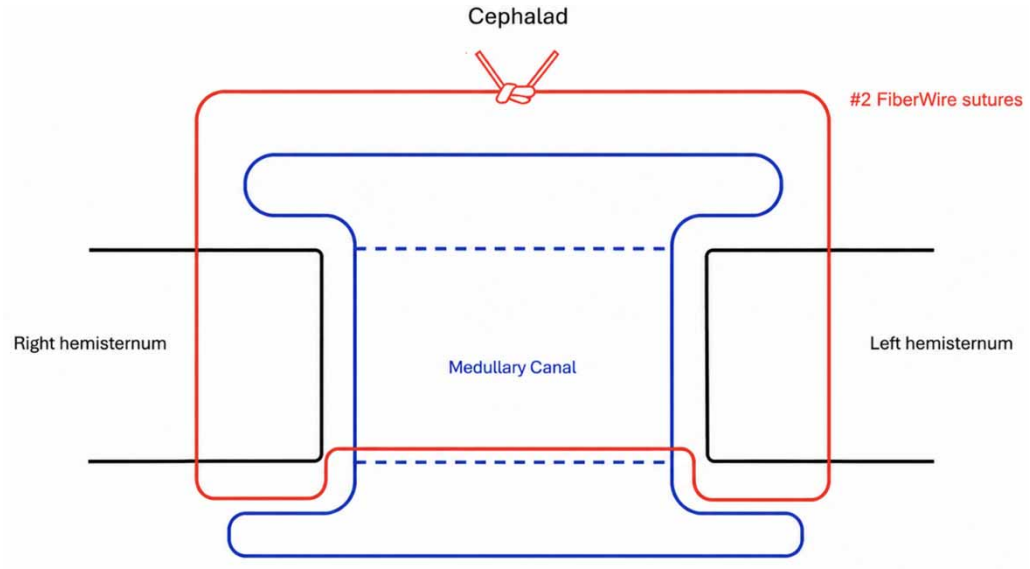


Figure 3 Schematic of fibular allograft strut fixation: the cross-sectional illustration demonstrates fixation of the distracted sternum using a custom-fashioned fibular allograft strut. The U-shaped ends of the graft are contoured to seat against the medial aspect of each hemisternum, providing geometric stability while maintaining the distracted position. Preloaded #2 FiberWire sutures are passed through the medullary canal of the graft and secured onto the cartilaginous sternum, allowing fixation without the need for cortical screw purchase.

specifically for SDS-associated thoracic insufficiency. The current construct also differs in its staged distraction and use of fibular allograft stabilisation in an infant whose sternum was insufficiently ossified for screw-based fixation.

The early physiological changes are encouraging but should be interpreted cautiously. Chest circumference increased from 36 to 41cm; peak inspiratory pressures decreased from between 40 and 50cm H_2O to the mid-20cm H_2O range at 3 months and remained in the upper 20cm range at 6 months; pulmonary hypertension was no longer evident on follow-up echocardiography; and inhaled nitric oxide was discontinued. The patient nevertheless remained ventilator dependent. Established pulmonary hypoplasia and prolonged preoperative ventilatory dependence may limit the speed and extent of respiratory recovery after mechanical thoracic expansion. These observations therefore demonstrate sustained early improvement in practical physiological surrogates, not proof of normalised lung function or long-term benefit.

Long-term management will focus on serial assessment of thoracic growth, graft position and incorporation, ventilatory requirements and pulmonary vascular status. Chest circumference and other thoracic measurements will be followed longitudinally, with interval radiography and computed tomography reserved for clinically indicated assessment of anatomy or graft incorporation. Serial echocardiography will be used to monitor for recurrent pulmonary hypertension. The skeletal phenotype in SDS evolves with age, but the extent of residual rib and thoracic growth in a severely affected infant is uncertain.[2] No specific future reconstruction is predetermined; VEPTR, lateral thoracic expansion or another age-appropriate reconstruction will be considered if recurrent thoracic restriction develops as the child grows. The current construct should therefore be viewed as an early rescue/bridge intervention, the durability and effects on subsequent thoracic growth of which require continued follow-up.

Conclusion

In carefully selected infants with severe thoracic insufficiency, staged sternal distraction may provide meaningful thoracic expansion when conventional reconstruction is not yet feasible. This case also highlights that severe thoracic dystrophy can occur as a rare manifestation of Shwachman-Diamond syndrome.

Competing interests

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Ethics approval and consent to participate

Ethics approval was not required for this single-patient case report in accordance with institutional policy. Written informed consent for publication was obtained from the patient/s parent.

Author contributions

A Jarrin Lopez: Writing – original draft. **A Zhao:** Formal analysis, Writing – review & editing. **R Darawsheh:** Data curation, Writing – review & editing. **D Kephart:** Methodology. **VA Sebastian:** Methodology. **O Idowu:** Investigation. **C Babbitt:** Formal analysis, Writing – review & editing. **S Kim:** Conceptualisation, Data curation, Formal analysis, Investigation, Methodology, Project administration, Supervision, Writing – review & editing.

Artificial Intelligence

The author/s declare that AI (ChatGPT) was used during manuscript preparation for language and grammar editing, to improve stylistic clarity, and to support the literature search and review process. All scientific content, data interpretation, literature selection, and final text were independently verified, reviewed, and finalised by the authors.

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